Hereditary Cancer Solution by SOPHiA GENETICS (Em

Assessing genetic predisposition to cancer for confident decision making

The first CE-IVD capture-based hereditary cancer solution on the market

The Hereditary Cancer Solution (HCS) by SOPHiA GENETICS is a CE-IVD marked molecular diagnostic application that bundles a capture-based target enrichment kit with the analytical power of SOPHiA™ AI and full access to SOPHiA DDM® platform.

The solution was expertly designed to accurately characterize the complex mutational landscape of the major hereditary cancer disorders such as breast and ovarian cancer, HNPCC and intestinal polyposis syndromes.

Highlights

- Relevant gene content covering breast, ovarian and digestive cancers
- Clinical grade performance
- Robust solution designed to detect complex variants such as long Indels, ALU insertions and differentiate
- High productivity workflow with short turnaround time







Smart Kit Design •

- · High affinity probe design ensuring unmatched coverage uniformity over the whole panel including complex genomic regions such as GC-rich regions
- Comprehensive panel of 26 genes carefully selected by experts from several leading institutions worldwide
- Automated workflow available on leading liquid handling robots for accurate, reliable, and high-throughput library preparation

- · Analytical performance matching diagnostic requirements (i.e. 99.99% of specificity and accuracy)
- High-confidence calling of SNVs, Indels and CNVs in all the genes of the panel
- Advanced detection of ALU insertions
- Efficient differentiation of PMS2 and PM2CL variants

Platform

• Intuitive and user-friendly interface

- Full control over accessibility with secure storage of anonymized data
- Dedicated features facilitating data visualization and interpretation
- Customized reporting including medical information and data results



Better diagnose to better care

SOPHiA GENETICS helps healthcare professionals to achieve better and faster diagnosis of patients worldwide. When choosing our solutions, clinicians benefit from:



Our universal technology, SOPHiA AI, supporting over 150 clinical grade genomic applications

Our Validation Program, facilitating rapid adoption of genomic applications in routine diagnostics and ISO 15189 accreditation

Our secure data processing and storage policy, complying with national and international guidelines

SOPHiA's community, allowing anonymized and safe knowledge sharing among experts worldwide